



DHCR7 gene

7-dehydrocholesterol reductase

Normal Function

The *DHCR7* gene provides instructions for making an enzyme called 7-dehydrocholesterol reductase. This enzyme is responsible for the final step in cholesterol production in many types of cells. Specifically, 7-dehydrocholesterol reductase converts a molecule called 7-dehydrocholesterol to cholesterol.

Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods that come from animals (particularly egg yolks, meat, poultry, fish, and dairy products). It has important functions both before and after birth. Cholesterol plays a critical role in embryonic development by interacting with signaling proteins that control early development of the brain, limbs, genital tract, and other structures. It is also a structural component of cell membranes and myelin, the fatty covering that insulates nerve cells. Additionally, cholesterol is used to make certain hormones and is important for the production of acids used in digestion (bile acids).

Health Conditions Related to Genetic Changes

Smith-Lemli-Opitz syndrome

More than 120 mutations that cause Smith-Lemli-Opitz syndrome have been identified in the *DHCR7* gene. The most common mutation, which is written as IVS8-1G>C, alters a single DNA building block (nucleotide) in the gene. This change interferes with the normal processing of 7-dehydrocholesterol reductase. Another common mutation occurs frequently in affected individuals of Mediterranean heritage. This mutation replaces one protein building block (amino acid), called threonine, with another amino acid, methionine, at position 93 in the enzyme (written as Thr93Met or T93M).

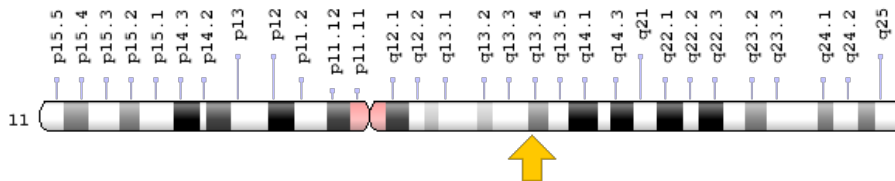
Most of the known *DHCR7* mutations change single amino acids in 7-dehydrocholesterol reductase. These mutations reduce the ability of this enzyme to convert 7-dehydrocholesterol to cholesterol. Other mutations insert or delete nucleotides in the *DHCR7* gene or lead to the production of an abnormally short enzyme; these mutations eliminate the activity of the enzyme. Without functional 7-dehydrocholesterol reductase, cells are unable to produce enough cholesterol. In addition, potentially toxic byproducts of cholesterol production (such as 7-dehydrocholesterol) can build up in the blood and other tissues. The combination of low cholesterol levels and an accumulation of related substances likely disrupts the growth and development of many body systems. It is not known, however, how this

disturbance in cholesterol production leads to the specific features of Smith-Lemli-Opitz syndrome.

Chromosomal Location

Cytogenetic Location: 11q13.4, which is the long (q) arm of chromosome 11 at position 13.4

Molecular Location: base pairs 71,434,411 to 71,448,431 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 7-DHC reductase
- D7SR
- delta-7-dehydrocholesterol reductase
- DHCR7_HUMAN
- sterol delta-7-reductase

Additional Information & Resources

GeneReviews

- Smith-Lemli-Opitz Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1143>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DHCR7%5BTIAB%5D%29+OR+%287-dehydrocholesterol+reductase%5BTIAB%5D%29%29+OR+%28%28sterol+delta-7-reductase%5BTIAB%5D%29+OR+%287-DHC+reductase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- 7-DEHYDROCHOLESTEROL REDUCTASE
<http://omim.org/entry/602858>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_DHCR7.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DHCR7%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2860
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1717>
- UniProt
<http://www.uniprot.org/uniprot/Q9UBM7>

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